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Sommario/riassunto	Rare diseases are a group of genetic disorders occurring in a small percentage of the population with the conditions being chronic but incurable. Approximately 7000 to 8000 different types have been identified and about 350 million people globally are affected in childhood and adulthood, resulting in enormous physical, mental, and psychological suffering and financial burden. It is imperative for medical scientists, clinicians, communities, and societies to ensure appropriate care is applied to ease the suffering of such patients. The extraordinary and unprecedented hallmark in the field of rare diseases has revolutionized modern human medicine with exciting and advancing developments of the genomic era over the last two decades. Patients with rare diseases have been receiving increasing benefits in care and life quality improvements than ever before. This book intends to share and exchange the advancing knowledge and experiences from the authors, who have the necessary expertise within the various topics and subjects in the research, diagnosis, and management of rare diseases. It is hoped they are able to provide further benefits to patients and families with the development of early and accurate diagnosis and effective therapies.